RAB23 gene

RAB23, member RAS oncogene family

Normal Function

The *RAB23* gene provides instructions for making a protein that is involved in a process called vesicle trafficking, which moves proteins and other molecules within cells in sac-like structures called vesicles. A vesicle forms when the cell membrane folds around a substance outside the cell (such as a protein). The vesicle is drawn into the cell, pinched off from the cell membrane (a process called endocytosis), and attached to the Rab23 protein. Once inside the cell, the vesicle is guided by the Rab23 protein to its proper destination. Vesicle trafficking is important for the transport of materials that are needed to trigger signaling during development.

Through the transport of certain proteins, the Rab23 protein regulates a specific developmental pathway called the hedgehog signaling pathway that is critical in cell growth (proliferation), cell specialization, and the normal shaping (patterning) of many parts of the body during embryonic development.

Health Conditions Related to Genetic Changes

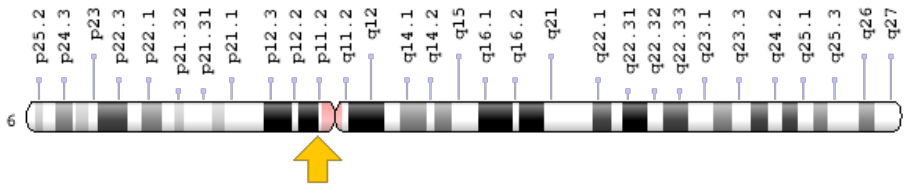
Carpenter syndrome

More than a dozen mutations in the *RAB23* gene have been found to cause Carpenter syndrome, a condition characterized by irregular skull formation, finger and toe abnormalities, and many other features. One mutation that is frequently seen in individuals with Carpenter syndrome who are of northern European ancestry replaces the protein building block (amino acid) leucine with a premature stop signal at protein position 145 (written as Leu145Term or L145X). This mutation results in an abnormally short, unstable protein that is quickly broken down. Other *RAB23* gene mutations that cause Carpenter syndrome reduce or eliminate function of the Rab23 protein. It is not clear how these mutations result in the specific features of Carpenter syndrome; however, it is likely that impaired transport of proteins involved in the hedgehog signaling pathway contributes to the development of this disorder.

Chromosomal Location

Cytogenetic Location: 6p12.1-p11.2, which is the short (p) arm of chromosome 6 between positions 12.1 and 11.2

Molecular Location: base pairs 57,186,992 to 57,222,314 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- HSPC137
- RAB family small GTP binding protein RAB 23
- RAB23_HUMAN
- ras-related protein Rab-23

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): The Hedgehog Pathway https://www.ncbi.nlm.nih.gov/books/NBK10043/#A1063
- Molecular Biology of the Cell (fourth edition, 2002): Rab Proteins Help Ensure the Specificity of Vesicle Docking https://www.ncbi.nlm.nih.gov/books/NBK26859/#A2325

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28RAB23%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- RAS-ASSOCIATED PROTEIN RAB23
  http://omim.org/entry/606144

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_RAB23.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RAB23%5Bgene%5D
- HGNC Gene Family: RAB, member RAS oncogene GTPases
  http://www.genenames.org/cgi-bin/genefamilies/set/388
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=14263
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/51715
- UniProt
  http://www.uniprot.org/uniprot/Q9ULC3

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